AUH gene

AU RNA binding methylglutaconyl-CoA hydratase

Normal Function

The *AUH* gene provides instructions for producing an enzyme called 3-methylglutaconyl-CoA hydratase. This enzyme is found in cell structures called mitochondria, which convert energy from food into a form that cells can use. Within mitochondria, this enzyme plays an important role in breaking down proteins into smaller molecules that cells can use to produce energy. Specifically, 3-methylglutaconyl-CoA hydratase is responsible for the fifth step in breaking down the protein building block (amino acid) leucine. The enzyme converts a molecule called 3-methylglutaconyl-CoA into another molecule called 3-hydroxy-3-methylglutaryl-CoA.

3-methylglutaconyl-CoA hydratase also has the ability to attach (bind) to RNA, a chemical cousin of DNA. Researchers are working to determine the purpose of this RNA-binding ability.

Health Conditions Related to Genetic Changes

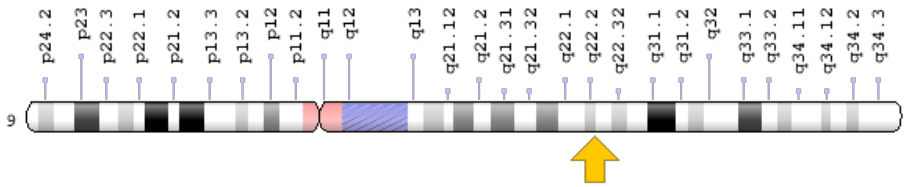
3-methylglutaconyl-CoA hydratase deficiency

At least 11 mutations in the *AUH* gene have been found to cause 3-methylglutaconyl-CoA hydratase deficiency. This condition causes neurological problems such as movement disorders and problems with thinking ability (cognition). The mutations that cause this condition lead to an absence of 3-methylglutaconyl-CoA hydratase enzyme activity. Without any functional 3-methylglutaconyl-CoA hydratase, the breakdown of leucine is incomplete. As a result, 3-methylglutaconyl-CoA is diverted into an alternative pathway and broken down into multiple acids: 3-methylglutaconic acid, 3-methylglutaric acid, and 3-hydroxyisovaleric acid. These acids accumulate in the body's fluids, causing elevated levels of acid in the blood (metabolic acidosis) and release of large amounts of these acids in urine (aciduria). Researchers speculate that an accumulation of these acids in the fluid that surrounds and protects the brain and spinal cord (the cerebrospinal fluid or CSF) can damage these structures and contribute to the neurological features of 3-methylglutaconyl-CoA hydratase deficiency.

Chromosomal Location

Cytogenetic Location: 9q22.31, which is the long (q) arm of chromosome 9 at position 22.31

Molecular Location: base pairs 91,213,815 to 91,361,969 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 3-methylglutaconyl-CoA hydratase
- 3-methylglutaconyl Coenzyme A hydratase
- AU RNA binding protein/enoyl-CoA hydratase
- AU RNA binding protein/enoyl-Coenzyme A hydratase
- AU RNA-binding protein/enoyl-Coenzyme A hydratase
- AU-specific RNA-binding protein
- AUMH_HUMAN
- enoyl-Coenzyme A hydratase
- methylglutaconyl-CoA hydratase

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): 3-Methylglutaconic Aciduria is Caused by Deficiencies of 3-methylglutaconyl-CoA hydratase, Which Mediates Formation of 3-hydroxy-3-methylglutaryl-CoA
  https://www.ncbi.nlm.nih.gov/books/NBK27945/#A3111
- Biochemistry (fifth edition, 2002): Inborn Errors of Metabolism Can Disrupt Amino Acid Degradation
  https://www.ncbi.nlm.nih.gov/books/NBK22493/
- Molecular Biology of the Cell (fourth edition, 2002): How Cells Obtain Energy from Food
  https://www.ncbi.nlm.nih.gov/books/NBK26882/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28AUH%5BTIAB%5D%29+OR+%283-methylglutaconic+aciduria+type+I%5BTIAB%5D%29%29+OR+%28%283-methylglutaconyl-CoA+hydratase%5BALL%5D%29+OR+%28Methylglutaconyl-CoA+hydratase%5BALL%5D%29%29+AND+%28Genes%5BMH%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- AU-SPECIFIC RNA-BINDING PROTEIN
  http://omim.org/entry/600529

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=AUH%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=890
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/549
- UniProt
  http://www.uniprot.org/uniprot/Q13825

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  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3249181/
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